GENES

ALLELES

GENE INTERACTIONS

Department of Biology,
Medical University - Sofia

Gregor Mendel

http://www.britannica.com/

- Gregor Johann Mendel was born Johann Mendel in Heinzendorf bei Odrau (now Hynčice, Czech Republic), 1822.
- Education - University of Olmoutz, (particularly in the subjects of physics and math) and University of Vienna.
- Mendel joined the Augustinian Monastery in Brno, and was given the name Gregor.
- At that time, the monastery was a cultural center for the region, and Mendel was immediately exposed to the research and teaching of its members, and also gained access to the monastery's extensive library and experimental facilities.
- In 1853, upon completing his studies at the University of Vienna, Mendel returned to the monastery in Brno and was given a teaching position at a secondary school, where he would stay for more than a decade. It was during this time that he began the experiments for which he is best known.

MENDELIAN GENETICS

- Genetics started with Gregor Mendel's experiments with pea plants in the 1860s. He discovered for first time the discrete nature of inheritance.

- In classical genetics, gene is the unit of heredity determining a trait. The alternative forms of a gene are called alleles (e.g. for purple and white petals). The genes of an organism make its genotype, while the traits make its phenotype.

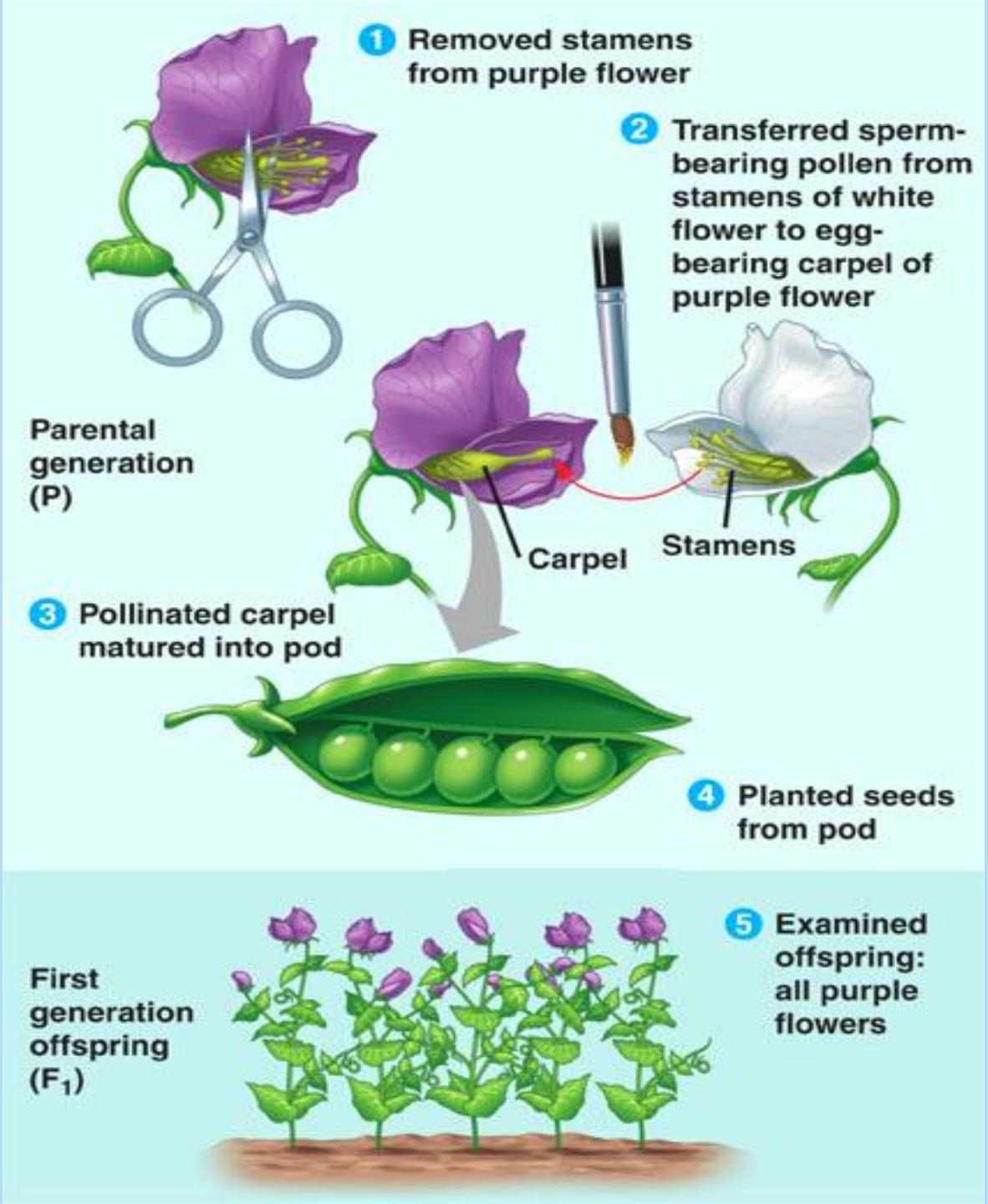
- A diploid eukaryote has every autosomal gene in 2 copies. When they are identical, the organism is homozygous for this gene. When they are different, the organism is heterozygous.

- Often, the heterozygote has the same phenotype as one of the homozygotes. The allele which is phenotypically expressed in this heterozygote is called dominant, and the "hidden" one is recessive. In Mendel's experiments, purple was the dominant trait and white was recessive.

Two phenotypes

The dominant trait
Photo: Mirna & Attilio Marzorati

The recessive trait
Photo: Burbuja Montoro



- Crossing pea plants
- That is how Mendel prevented self-fertilization.

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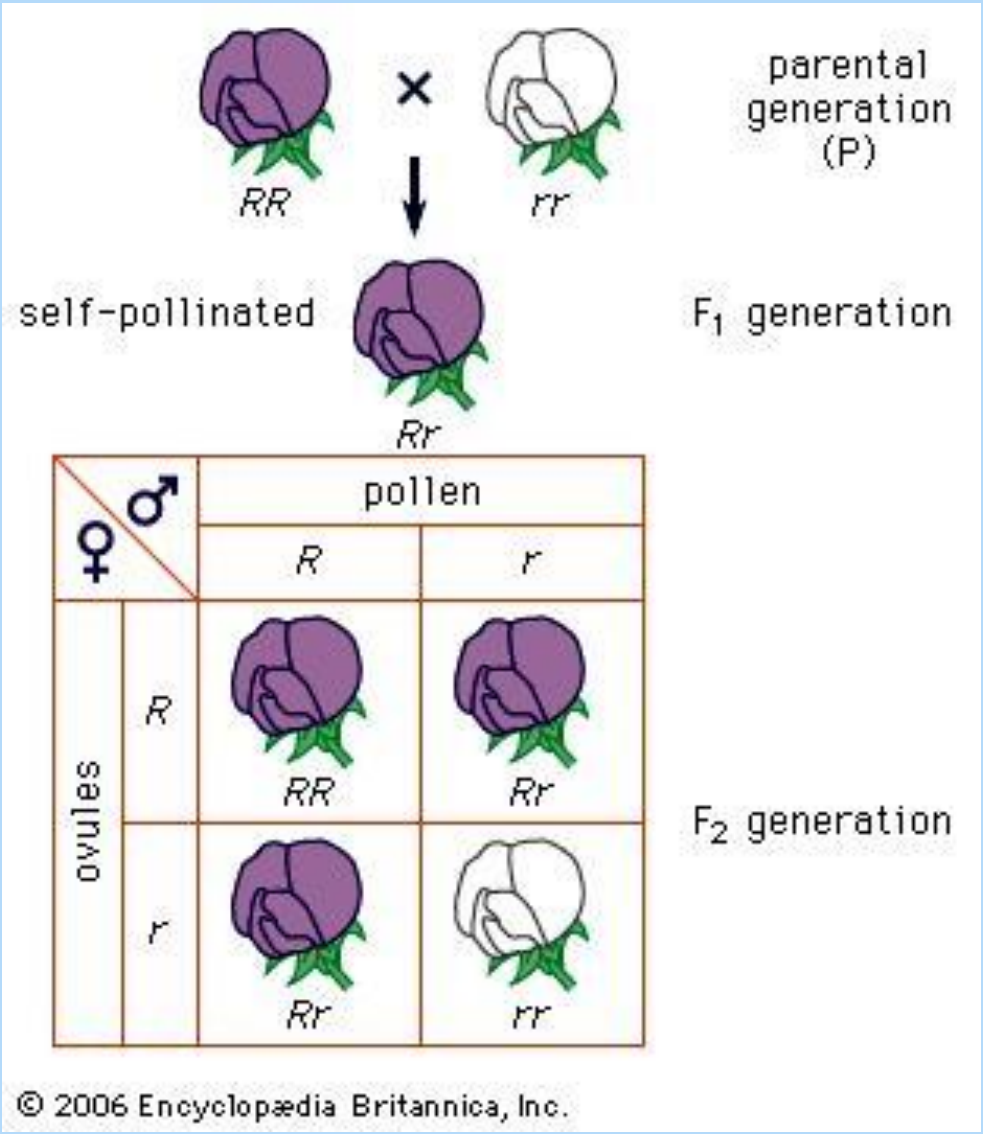
Mendel's law of segregation



- Cross of a purple-flowered and a white-flowered strain of peas.
- R stands for the gene for purple flowers
- r for the gene for white flowers.
- The law: in F1 all individuals have the dominant phenotype; in F2 the phenotypes are segregated in a relation 3:1 (dominant:recessive).

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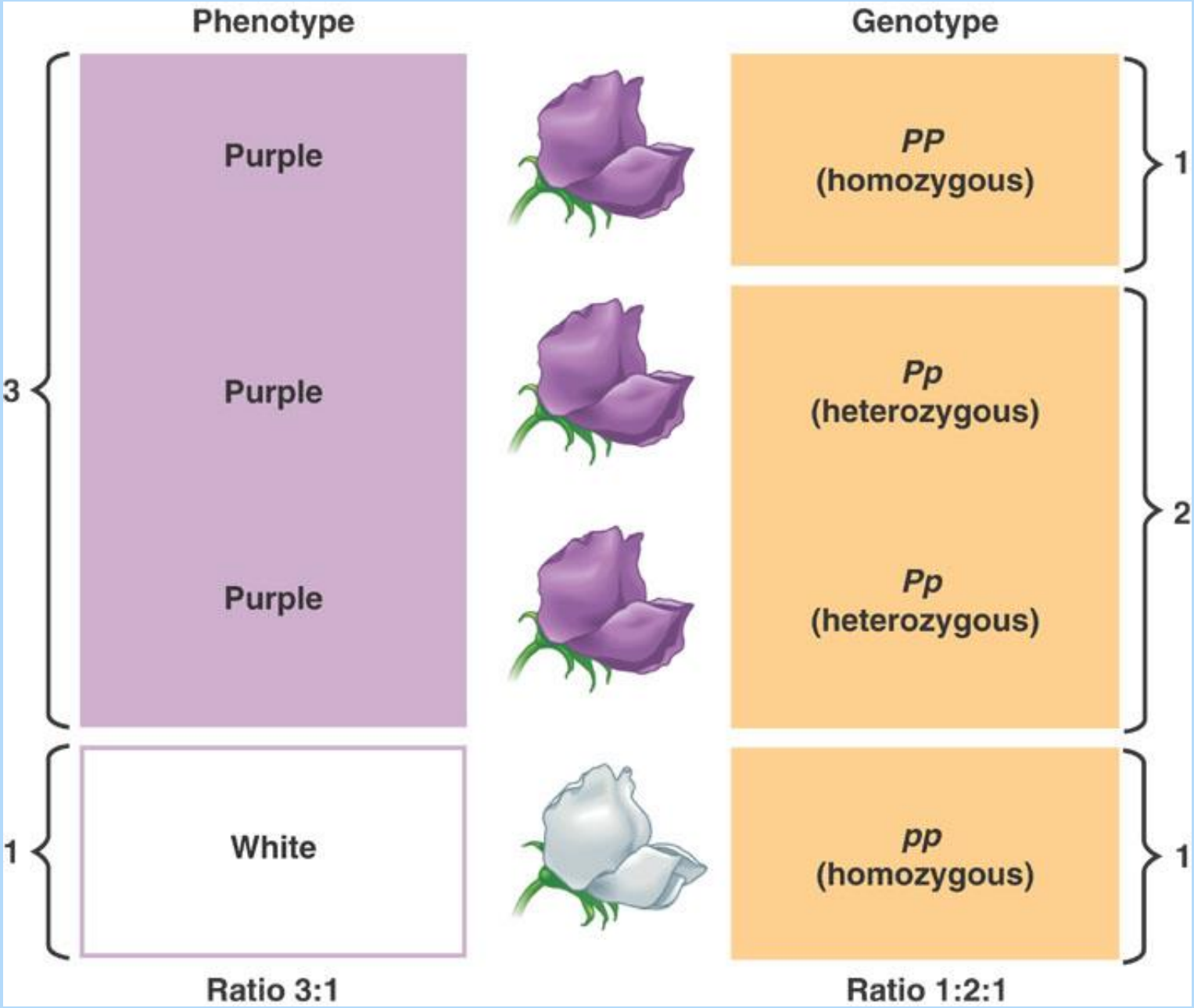
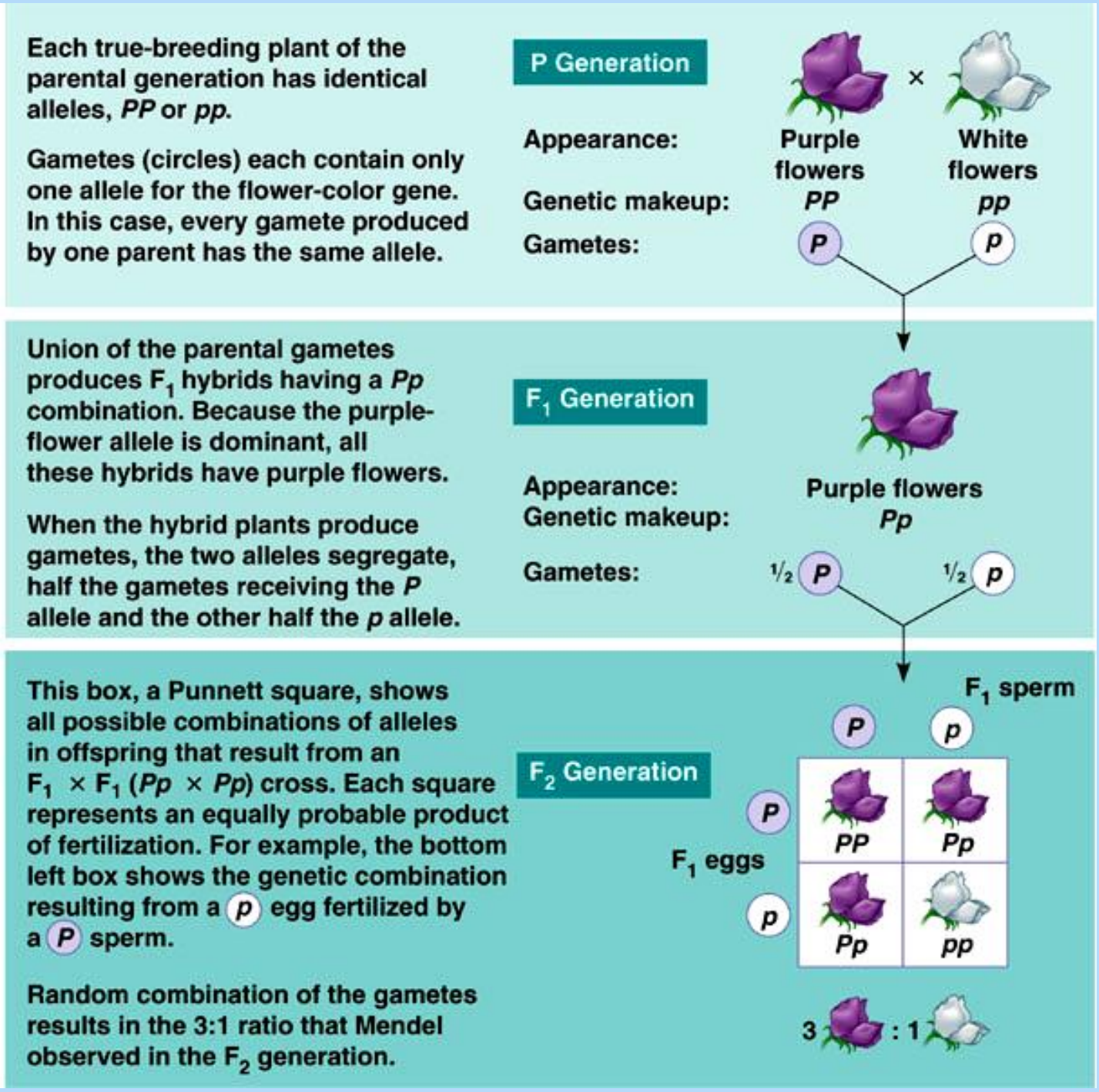


Phenotype
vs
Genotype



Law of segregation - explanation

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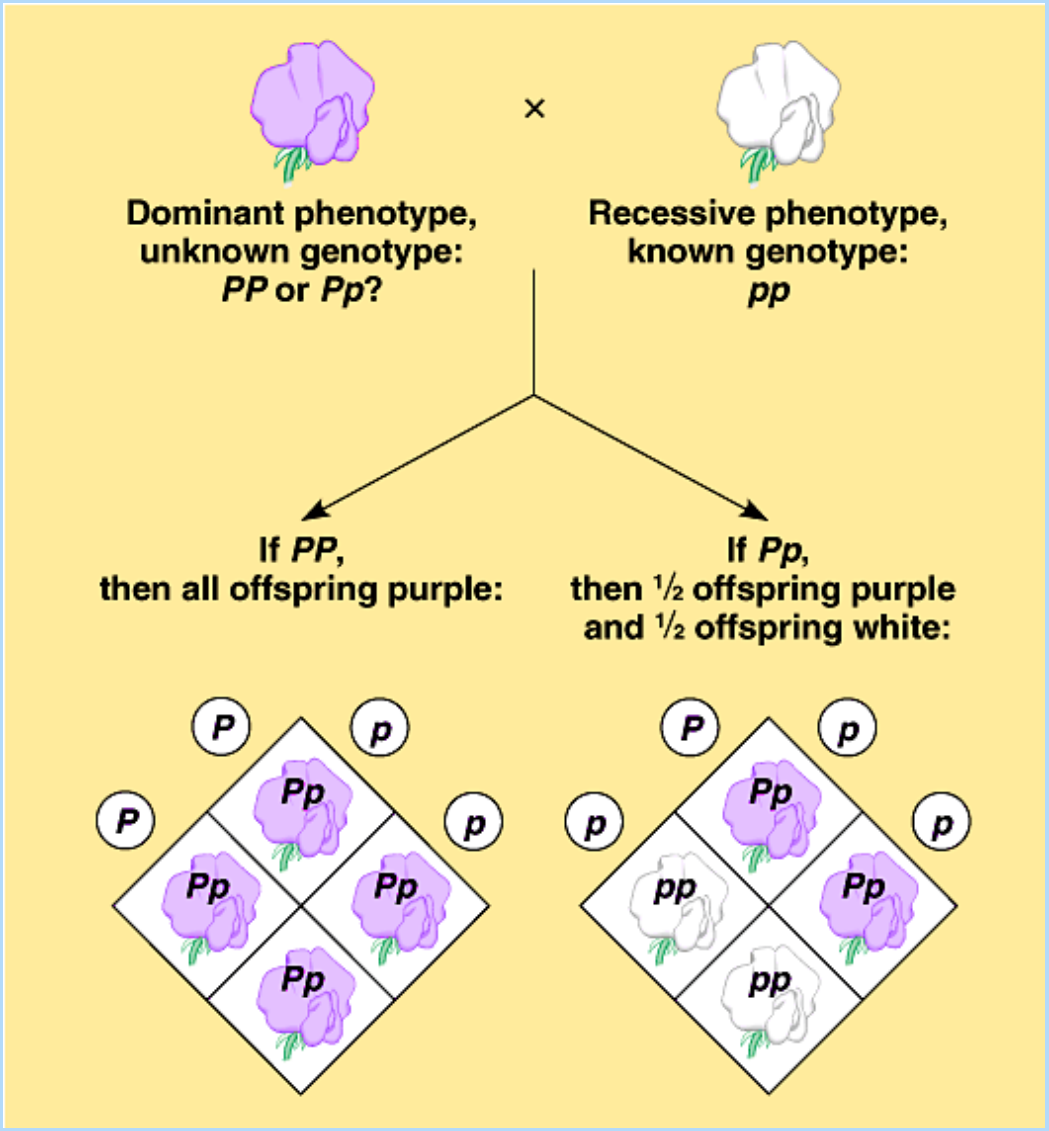
Types of crosses

- The dominant allele – by capital letter (e.g. A), the recessive allele – by the same small letter (e.g. a). Parents – by P, first generation – by F1, 2nd generation – F2 etc., cross – by x.
- When we trace the inheritance of 1 pair of alleles, the cross is called monohybrid. Two pairs – dihybrid, 3 pairs – trihybrid.
- Crossing reveals not only the type of inheritance of traits but also the genotype of particular individuals. If we want to know the genotype of an individual having the dominant trait (homo- or heterozygous), we cross it with an individual having the recessive trait (recessive homozygote). This is called test cross.

The test cross

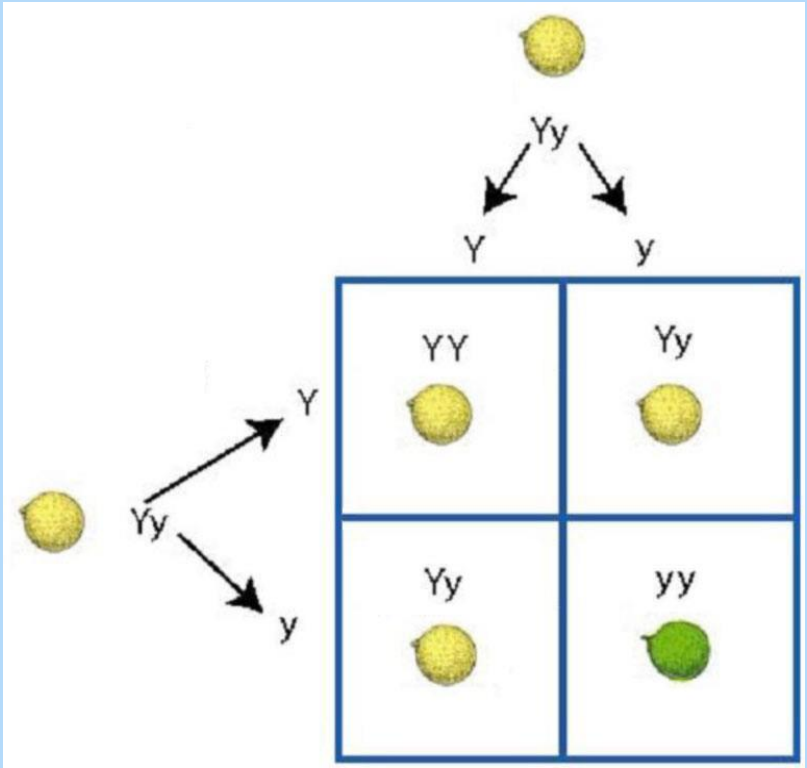
100% heterozygous organisms

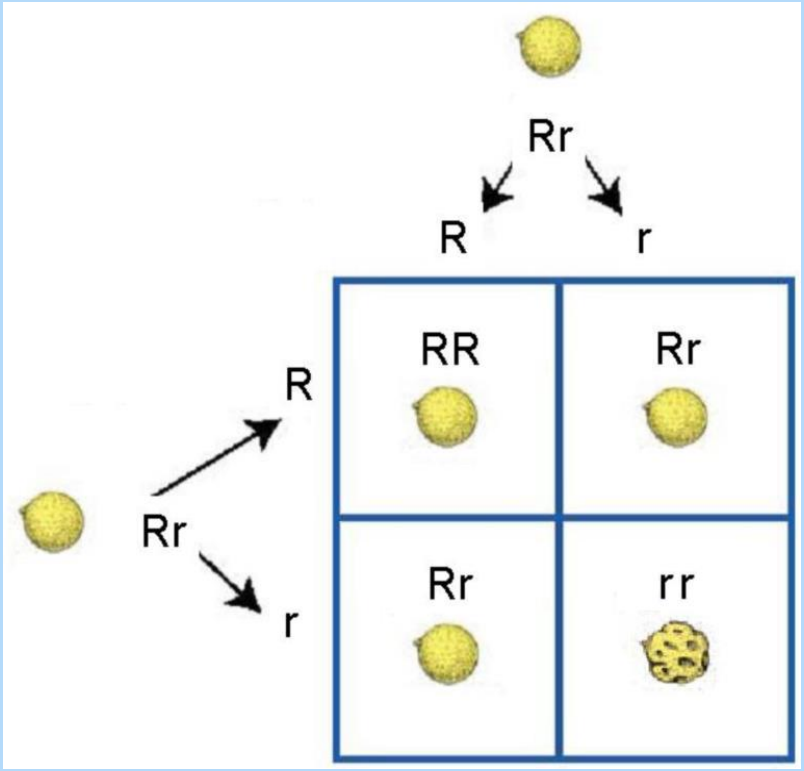


1 : 1 heterozygous and homozygous organisms

www.bio.utexas.edu

Other examples in Mendel's study



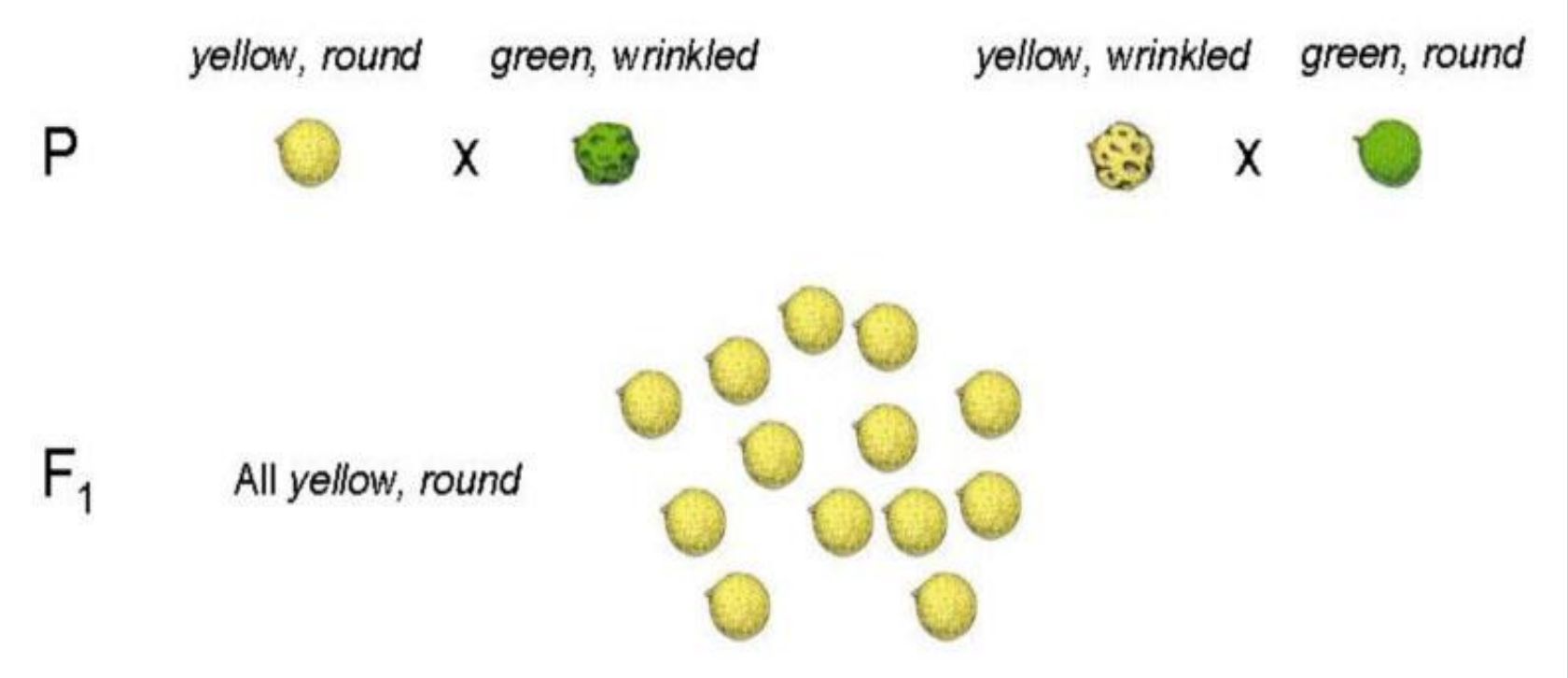


Yellow vs. green seed colour

Round vs. wrinkled seed surface

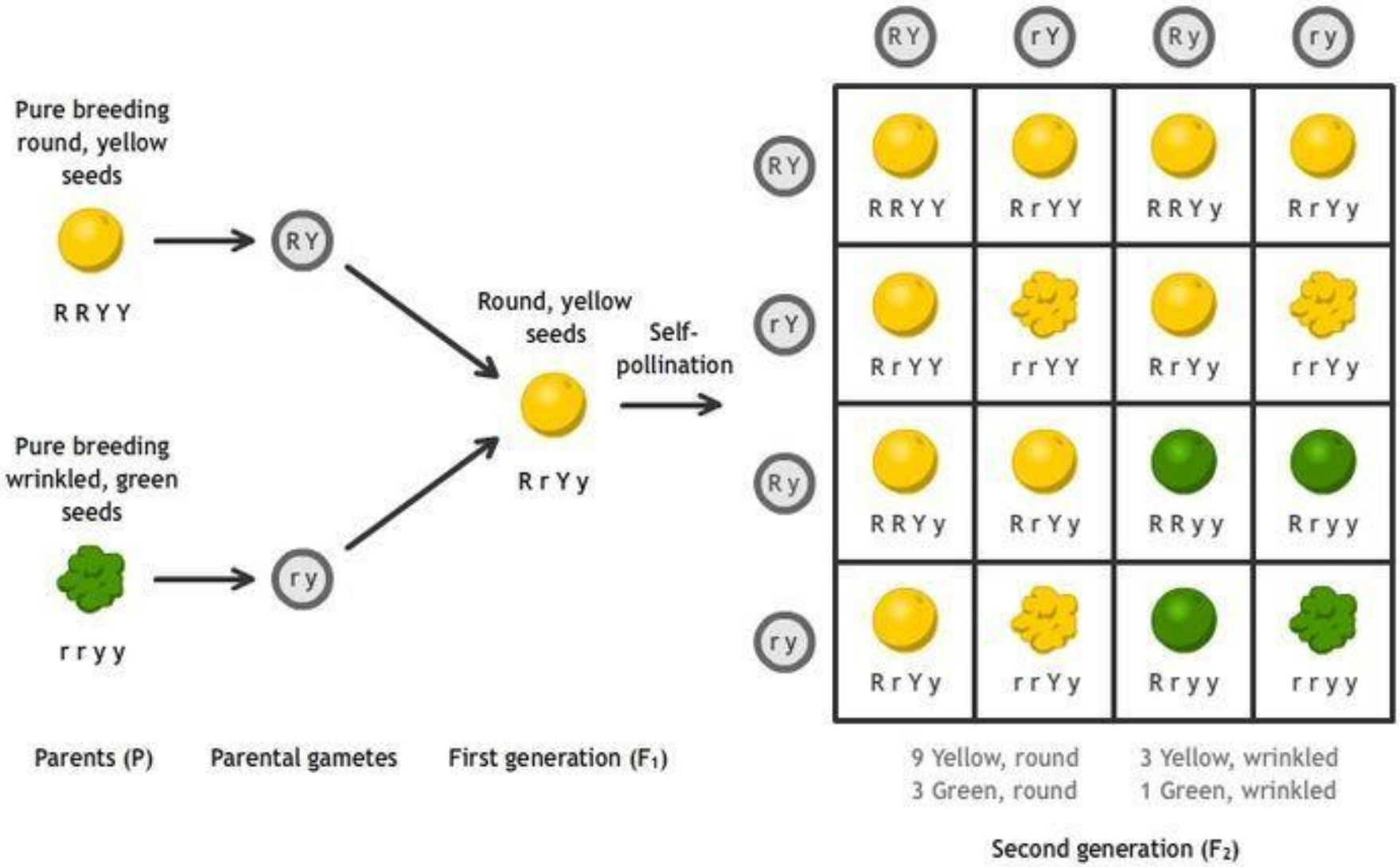
http://ib.berkeley.edu

Dihybrid cross



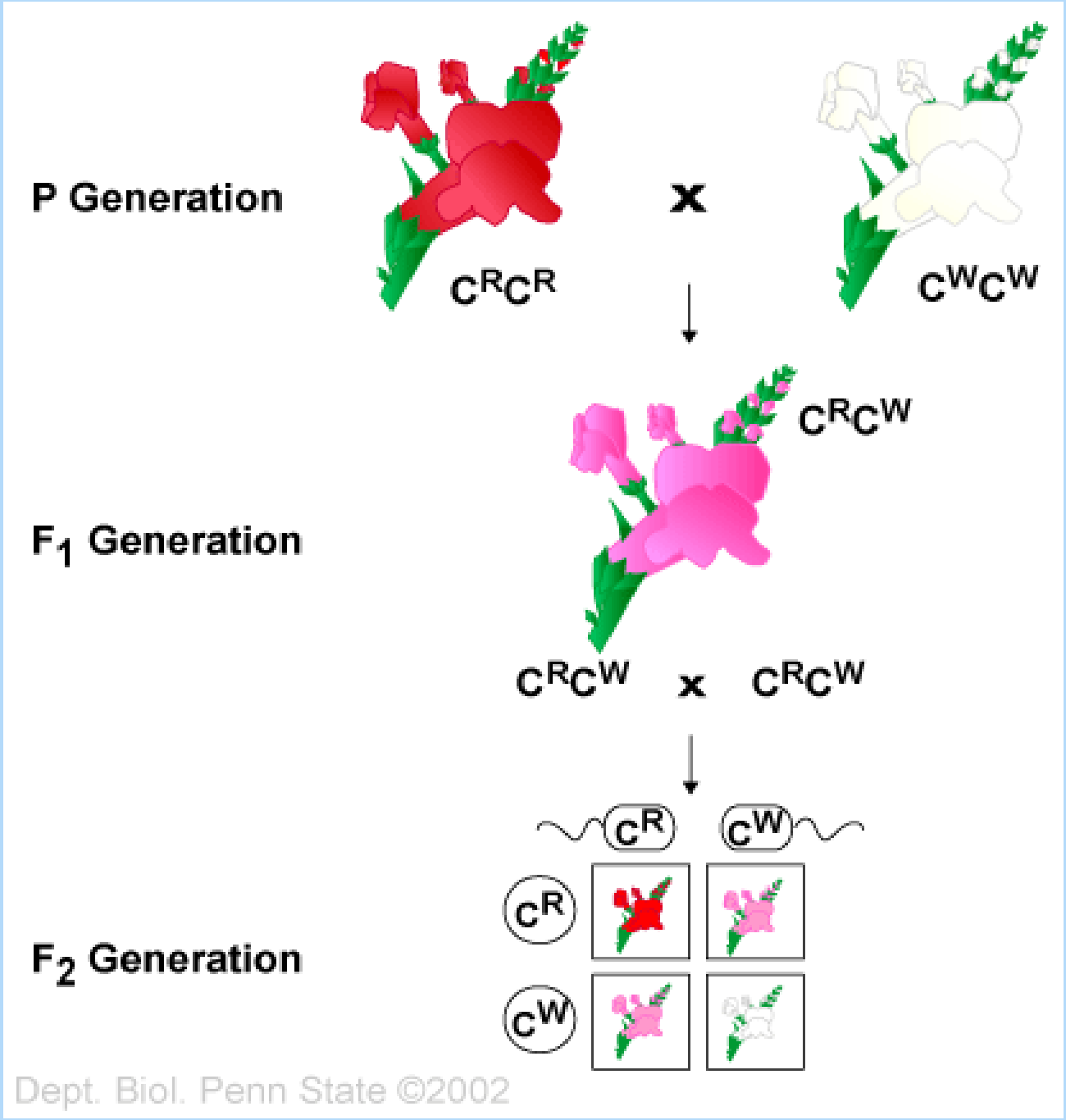
(from http://ib.berkeley.edu)

Phenotypes in F2 hybrids



Incomplete dominance

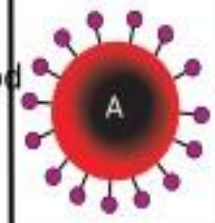
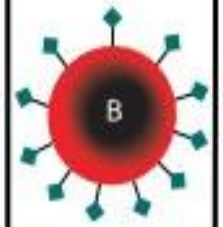
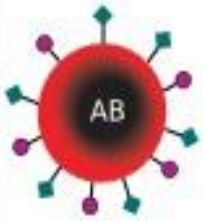
Some alleles show incomplete dominance. Heterozygotes have a phenotype intermediate to that of the two homozygotes. In F2, we have all 3 phenotypes 1:2:1.



Blood Type	Genotype		Can Receive Blood From:
A	$i^A i$ $i^A i^A$	AA AO	A or O
B	$i^B i$ $i^B i^B$	BB BO	B or O
AB	$i^A i^B$	AB	A, B, AB, O
O	$i\,i$	OO	O

Codominance

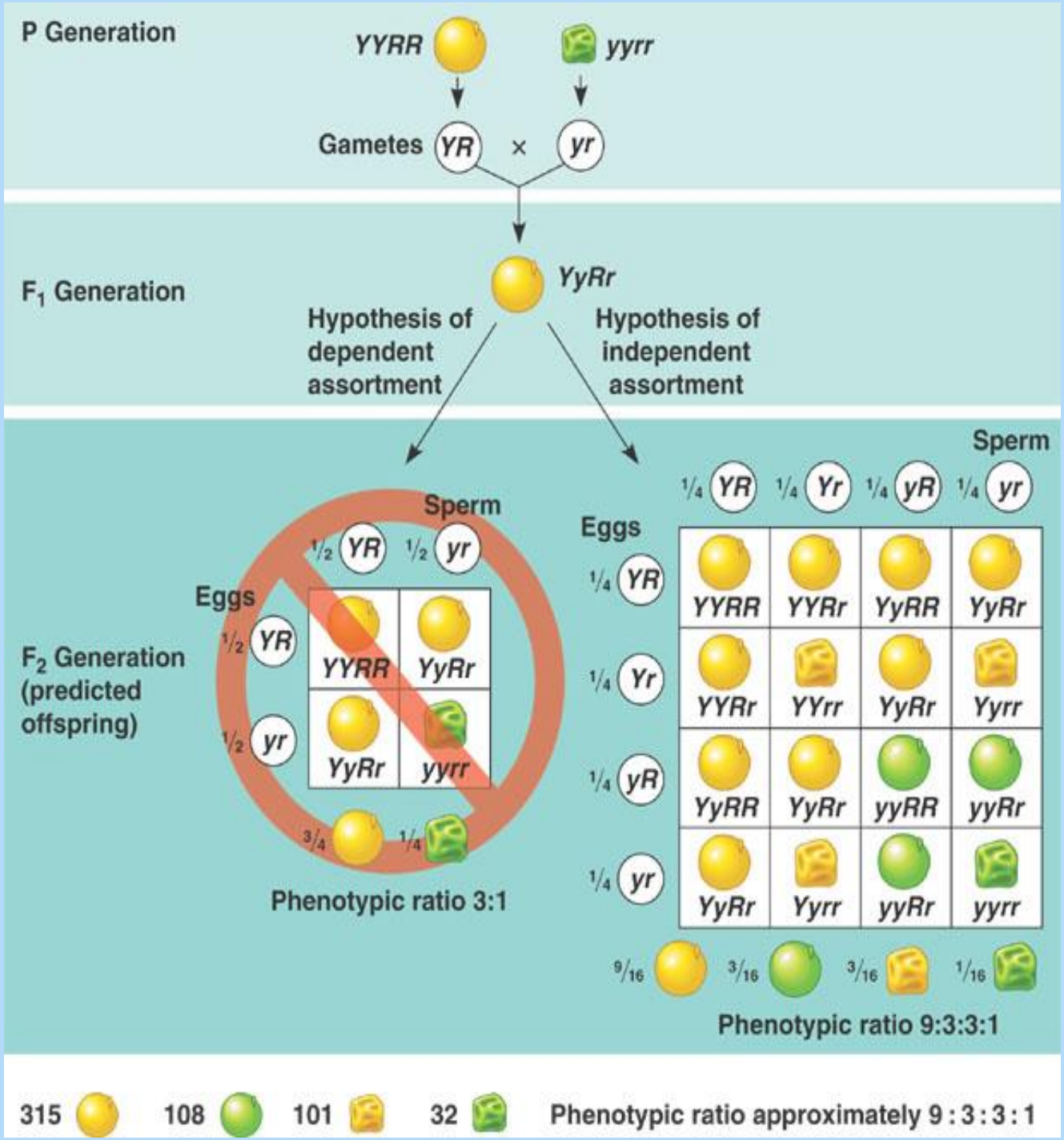
In codominant inheritance, the heterozygote expresses the traits of both alleles. E.g. the heterozygote for the A and B alleles from the ABO blood group system has phenotype AB.

	Group A	Group B	Group AB	Group O
Red blood cell type				
Antibodies present	 Anti-B	 Anti-A	None	 Anti-A and Anti-B
Antigens present	A antigen	B antigen	 A and B antigens	No antigens

Multiple alleles

The ABO blood group system is also an example of multiple allelism, i.e. more than 2 alleles in the same gene locus. Besides the A and B alleles, it has an O allele which is recessive to both A and B.

en.wikipedia.org



Independent assortment

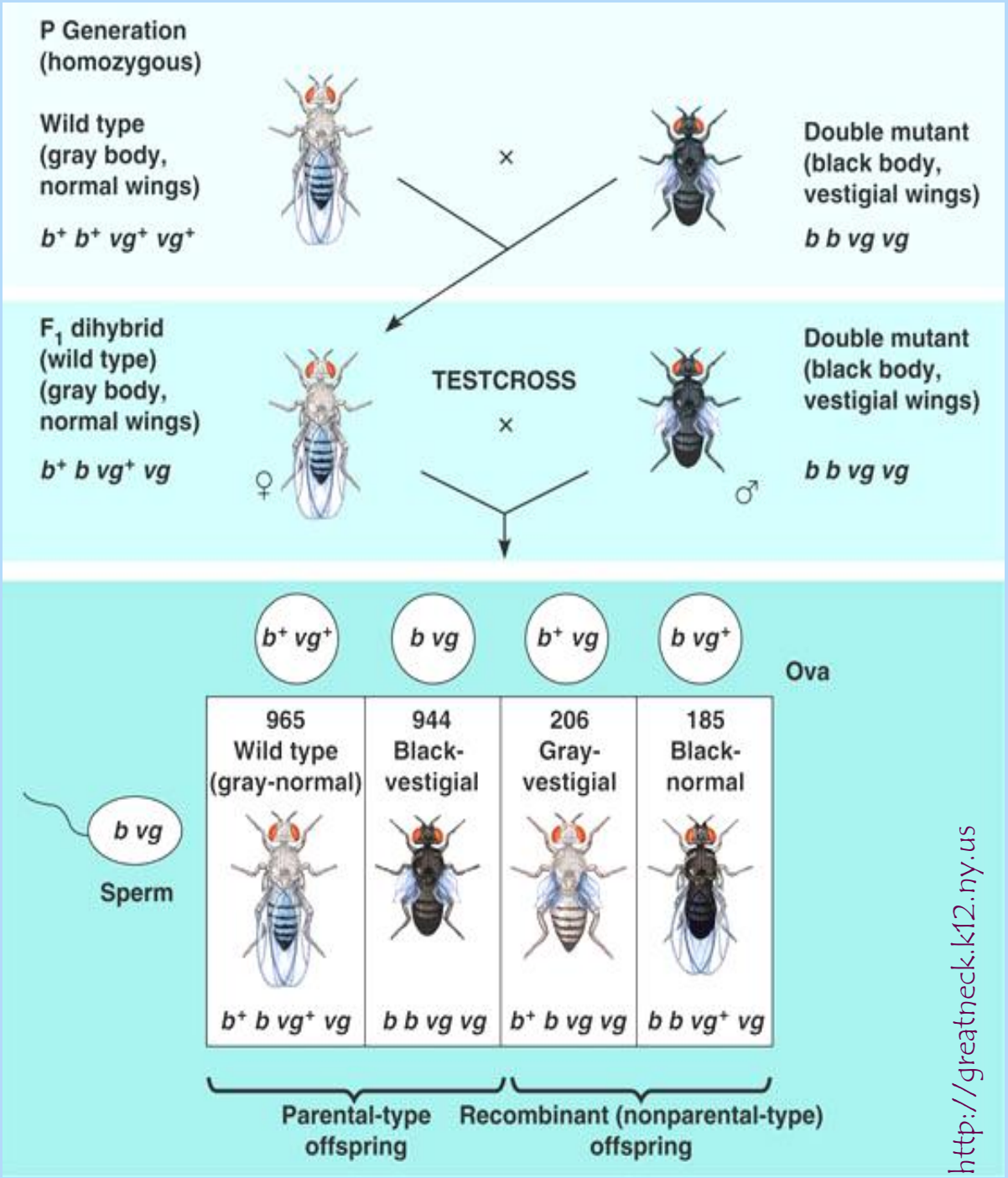
Mendel's law of independent assortment:

- Allele pairs separate independently during the formation of gametes.
- This means that traits are transmitted to offspring independently of one another.

en.wikipedia.org

- Thomas Hunt Morgan, (1866 - 1945) is an American zoologist and geneticist.
- He is famous for his experimental research with the fruit fly (*Drosophila*) by which he established the chromosome theory of heredity.
- He showed that genes are linked in a series on chromosomes
- Morgan's work played a key role in establishing the field of genetics. He received the Nobel Prize for Physiology or Medicine in 1933.

Linked genes



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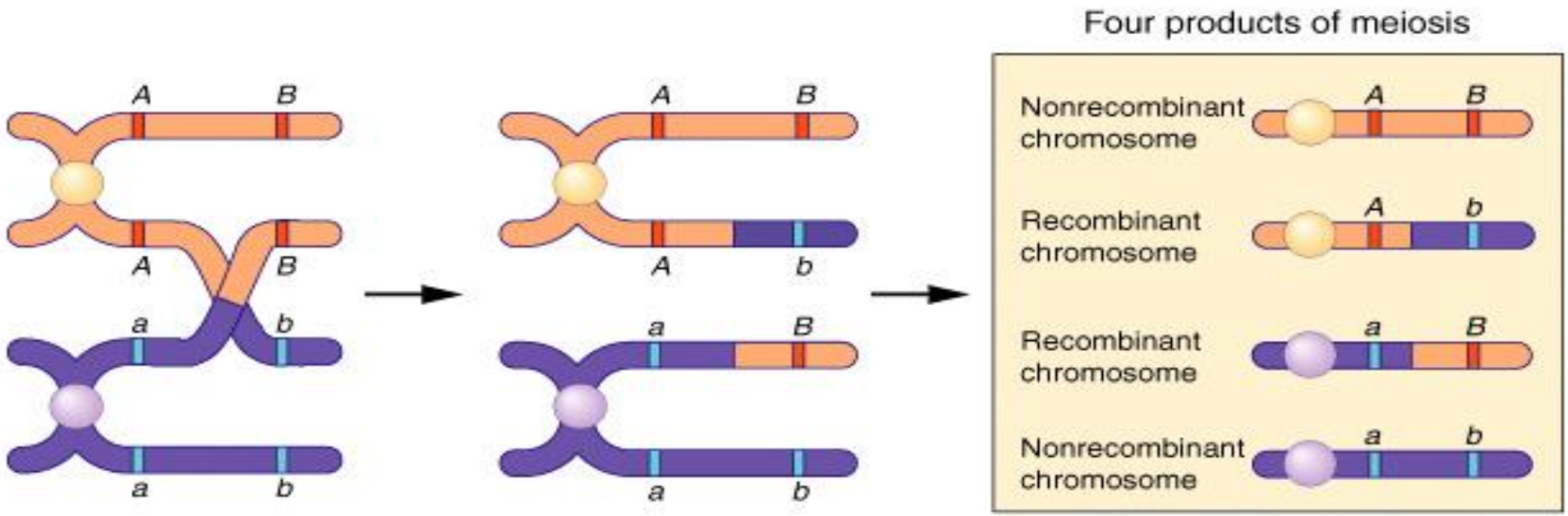
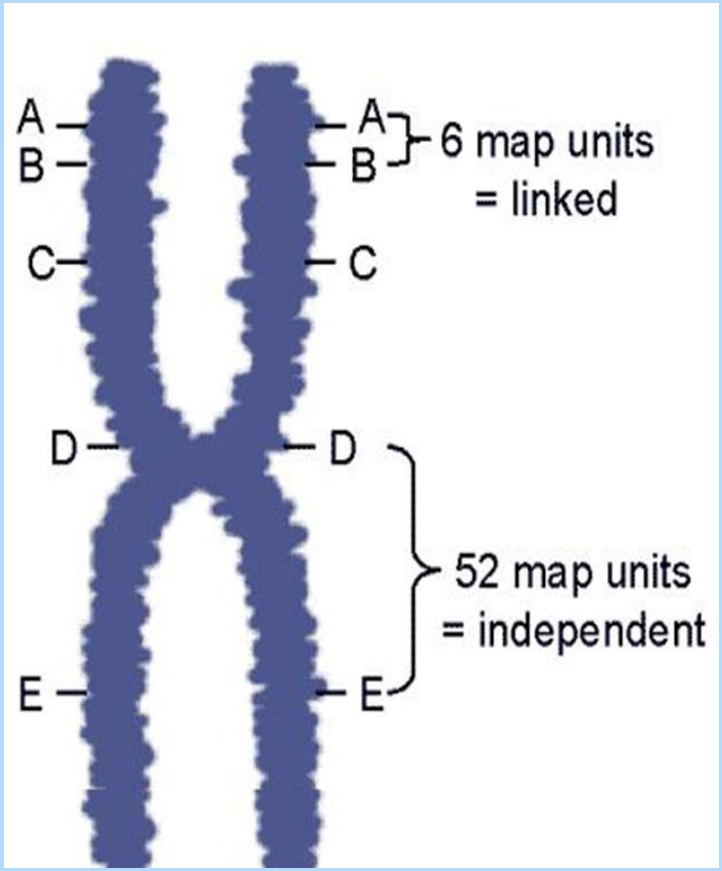
- Linked genes are inherited together with the other gene(s).
- When a pair or set of genes are on the same chromosome, they are usually inherited together or as a single unit.
- For example, in fruit flies the genes for eye color and the genes for wing length are on the same chromosome, thus are inherited together.
- Don't confuse them with sex-linked gene, which refers to a gene located on one of the sex chromosomes).

- Crossing over breaks off the gene linkage, because it changes the place of one allele from one to the other homologous chromosome.
- As a result – allele combinations in the gametes are different from those in somatic cells.
- The frequency of recombination between two genes depends on the distance between them. This is because crossing over on one place interferes occurring of other crossing over in the close vicinity. Genes which are closer in the chromosomes are often inherited together. Crossing over can not break their linkage.
- By the frequency of recombination between the genes of the fruit fly, Morgan makes maps of the Drosophila`s chromosomes.



- Gene linkage disturbs independent assortment of the genes (one of the requirements for Mendelian inheritance).
- Closely located genes in the chromosome is more likely to pass together to the offspring.
- If genes are far apart (or on different chromosomes), they are unlinked and assort independently.
- Linkage is measured by the recombination frequencies.
- According to some origins, only genes less than 50 map units apart are considered linked.



Sex determination

Chromosomal sex determination

XX/XY sex-determination system

(in most of mammals and some insects like Drosophila): females have two of the same kind

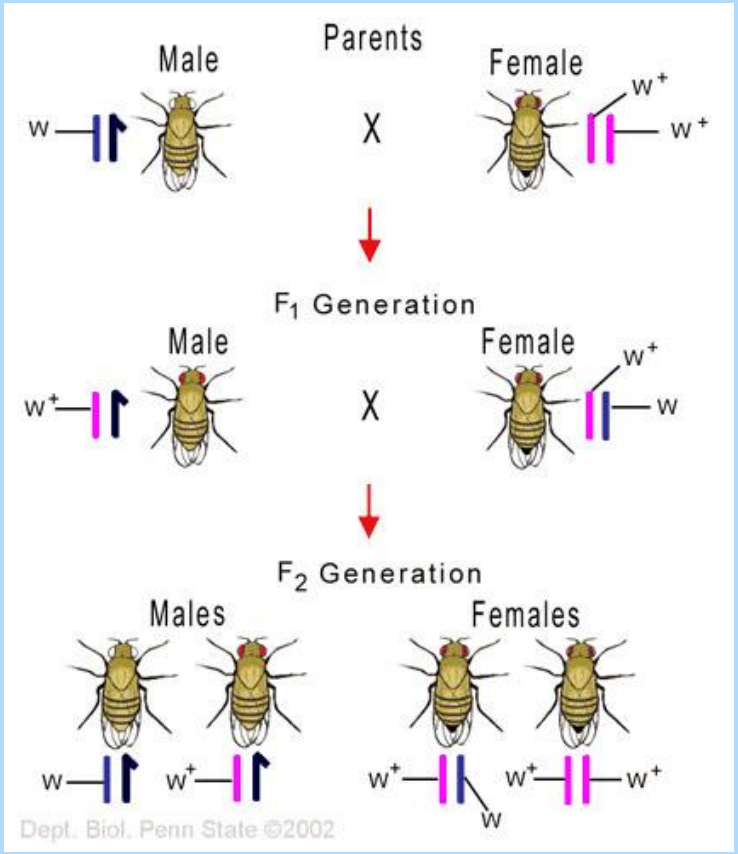
of sex chromosome (XX), while males have two distinct sex chromosomes (XY).

X and Y chromosomes are different in shape and size from each other, unlike the autosomes.

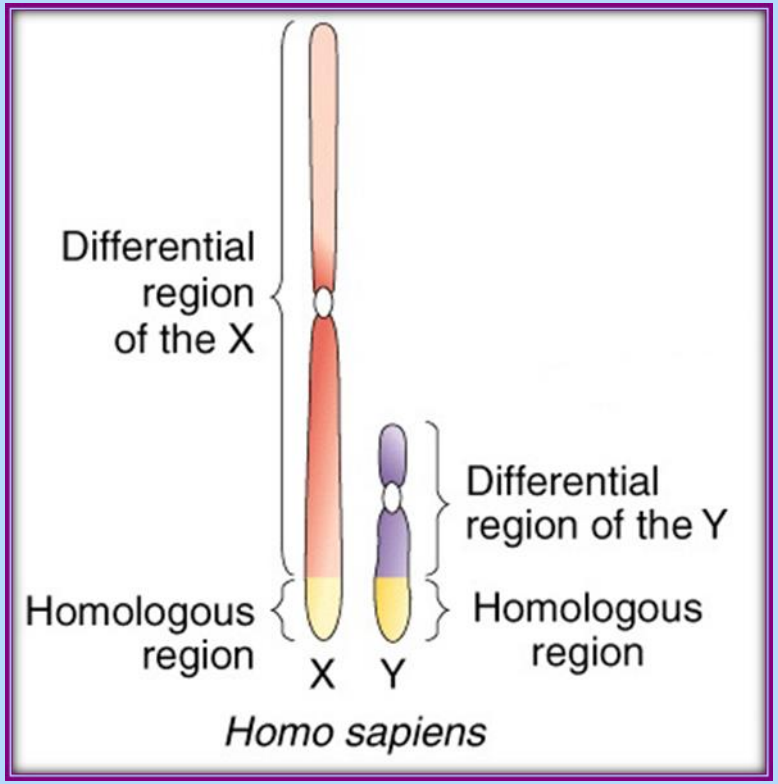
Mammal Y chromosome has a gene SRY that determines maleness.

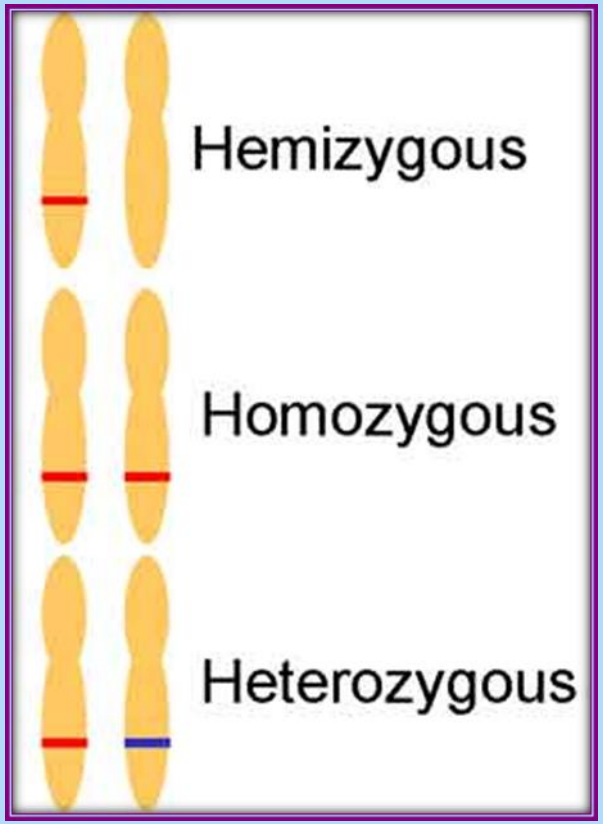
Sex-linked inheritance

- Sex-linked gene is a gene located on one of the sex chromosomes.
- For the gene in the X chromosome males are hemizygous. They inherit X chromosome only from their mother. Females inherit X chromosome from both of parents. Because of that, females can be homozygous or heterozygous for the X-linked traits.
- Form their father boys inherit Y chromosome. That's why if we talk about Y-linked inheritance, it is only between a father and a son.
- Morgan observed that inheritance of eye color in Drosophila depends on the sex of the individuals.
- The gene is X-linked, red eyes are the dominant "wild type" trait, white eyes are recessive mutant trait.
- Here, just males have white eyes.



Hemizygosity

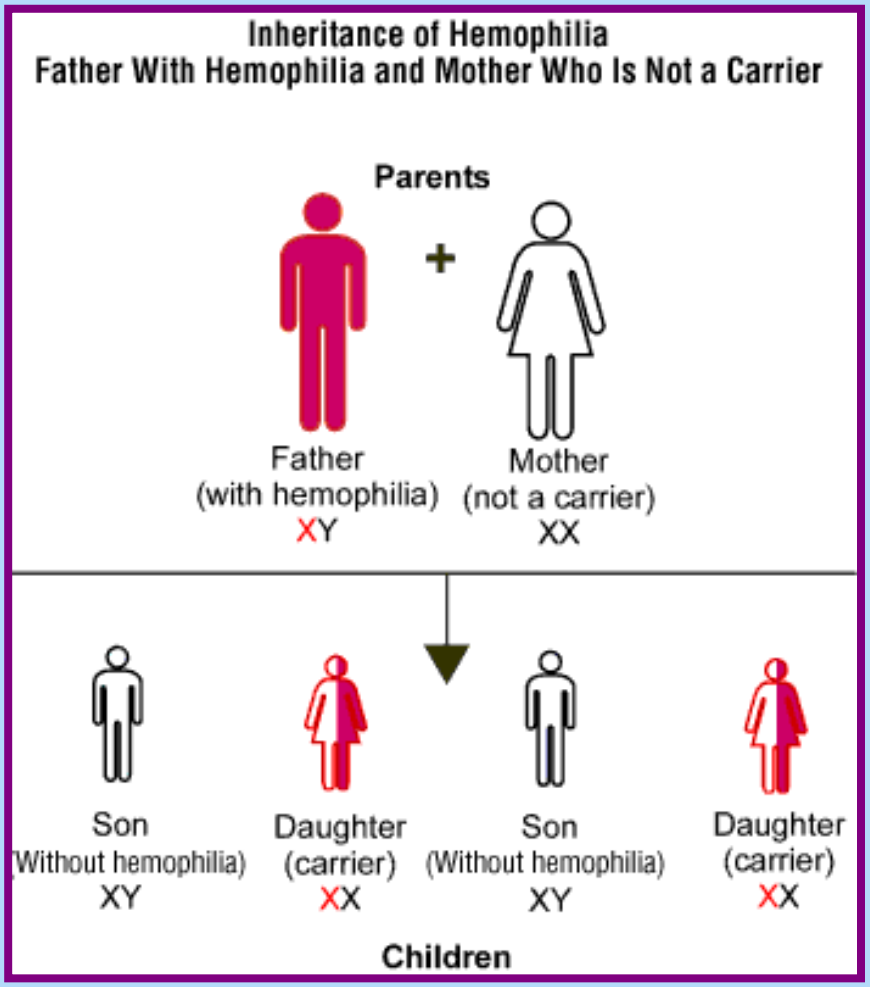


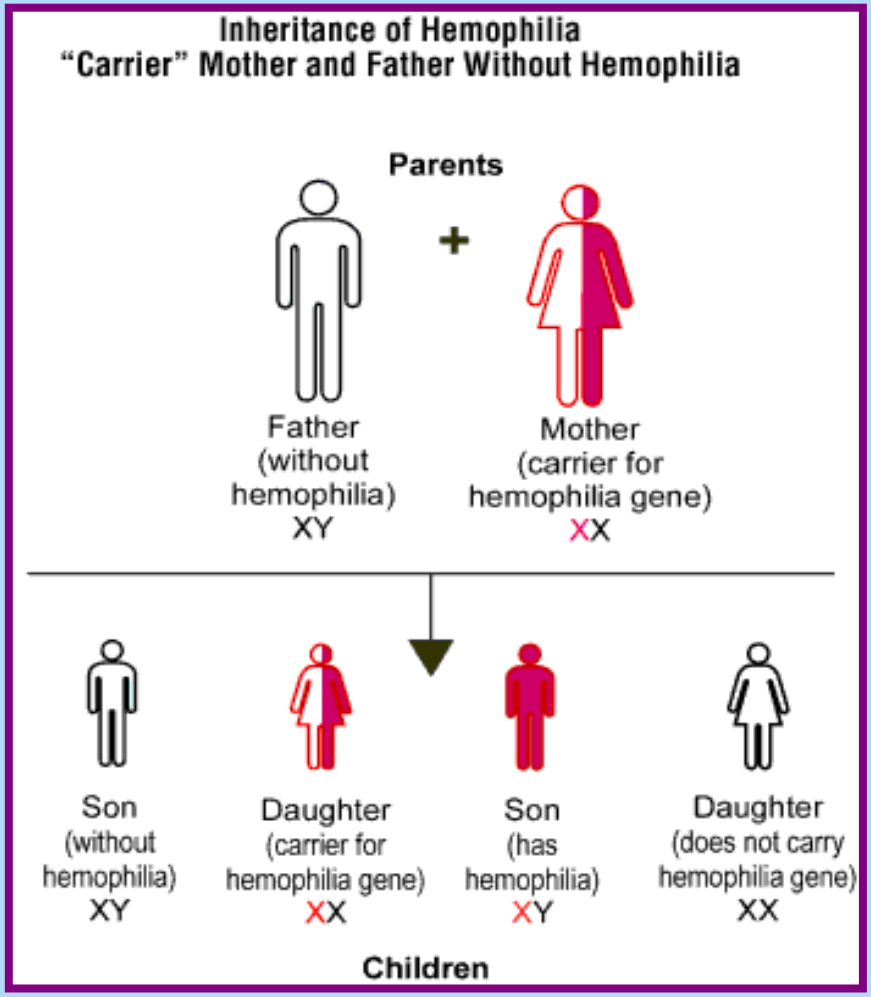


- Hemizygosity means a diploid cell to possesses only one allele of a gene instead of two.
- It could be a normal state like the sex-linked genes, but is possible to be abnormality too – if one autosomal allele is missing.
- For the X-linked genes male individuals (XY) are hemizygous.

SEX AND INHERITANCE

Sex-linked traits





medacad.wikispaces.com

Haemophilia in human

Another X-linked recessive condition is red-green colorblindness (Daltonism) in human

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Pedigree diagrams are used to trace inheritance in families